

FIGURE 1

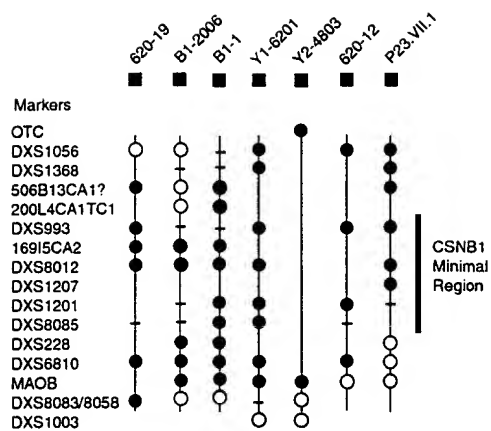
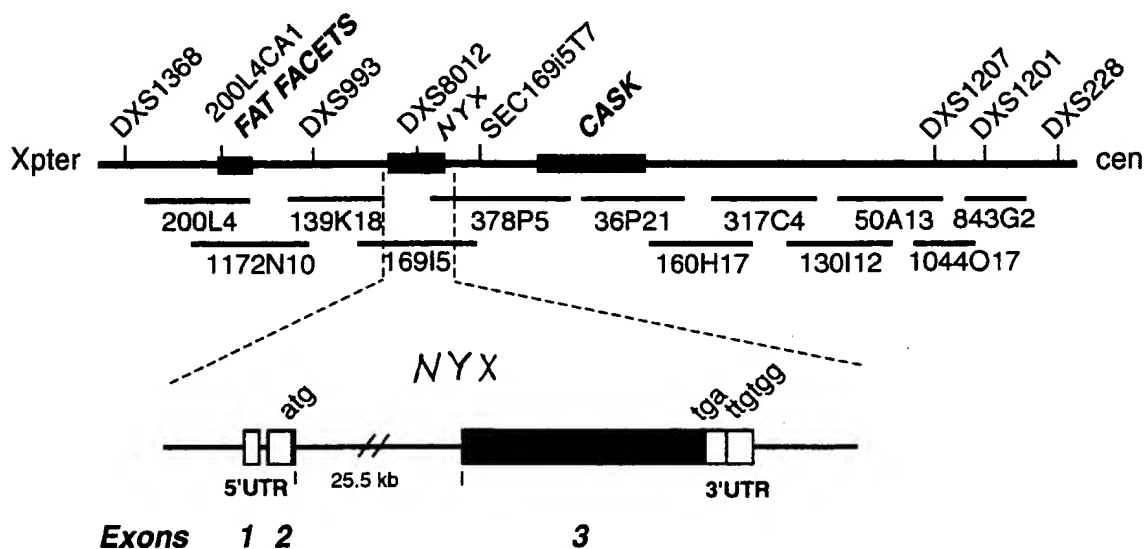


FIGURE 2

**a** Physical map of the CSNB1 minimal region and genomic organization of *NYX*



**b** Leucine-rich repeats and the distribution of mutations in the Nyctalopin protein

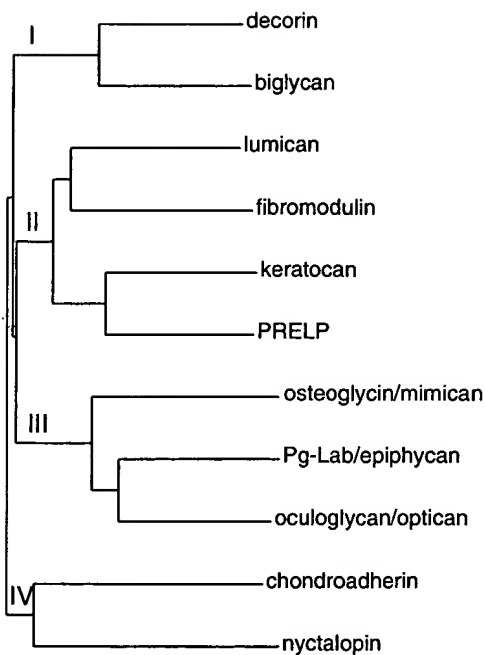
	MKGRGMLVLLHAAVVLGLPSAWAVGA <sup>✓</sup> ARA <u>PAA</u> AAESTVERGTSVRIDR	- 50
	<i>del</i>	
1.	AGLLRVPaelPCEAVSIDLDRNGL	- 74
2.	RFLGERAFGTLPSLRRRLSLRHNNL	- 98
3.	SFITPGAFKGLPRLAELRLAHNGDL	- 123
4.	RYLHARTFAALSRLRRRLDLAACRL	- 147
5.	F <sup>Λ</sup> SVPERLLAELPALRELAADFNLFRRVPGALRGL	- 181
6.	ANLTHAHL15LRRLRSLSLQANRV	- 218
	ERGRIEAVASSSLQ <sup>Λ</sup> G	
7.	RAVHAGAFGDCGVLEHLLLNNDLL	- 242
8.	AELPADAFRGLRRRLRTLNLGGAAL	- 266
9.	DRVARAWFADLAELELLYLDRNSI	- 290
10.	AFVEEGA <sup>Λ</sup> QNL SGLLALHLNGNRL	- 314
11.	TVLAWVAFQPGFFLGRLEFLFRNPW	- 338
	β-sheet                      α-helix	
	CCDCRLEWLRDWMEGSGRVTDVPASPGSVAGLDLSQVTFGRSSDGLQVD	- 388
	PEELNLTSSPGSPSEPAATTVSREFSLLSKLLAPRPVEEAANTTGGLA	- 438
	NASLSDSLSSRGVGGAGRQPWFLLASCLLPVAQHVVFGFLQMD	- 481

Λ insertions of SVPERLL, GLR and RLR, respectively  
 ✓ most likely signal peptide cleavage site

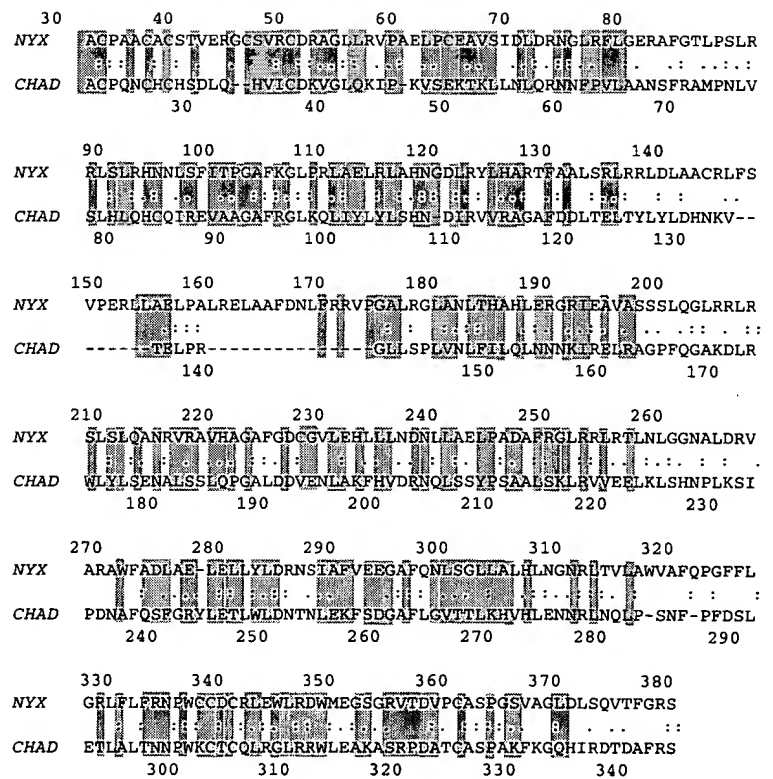
FIGURE 3

FIGURE 3  
CONT.

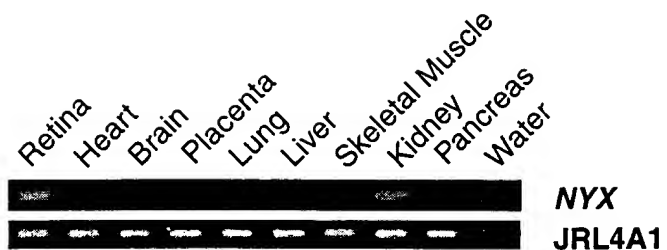
C



D

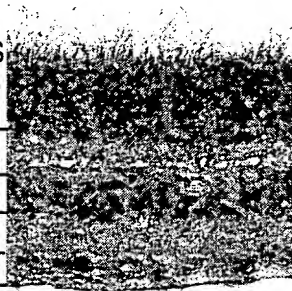


*a*



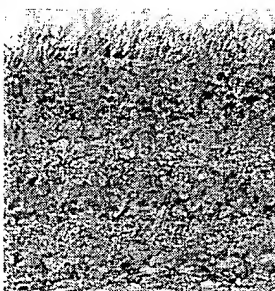
*b*

OS  
IS  
ONL  
OPL  
INL  
IPL  
GCL



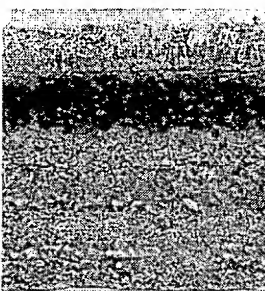
**NYX-AS**

*c*



**NYX-S**

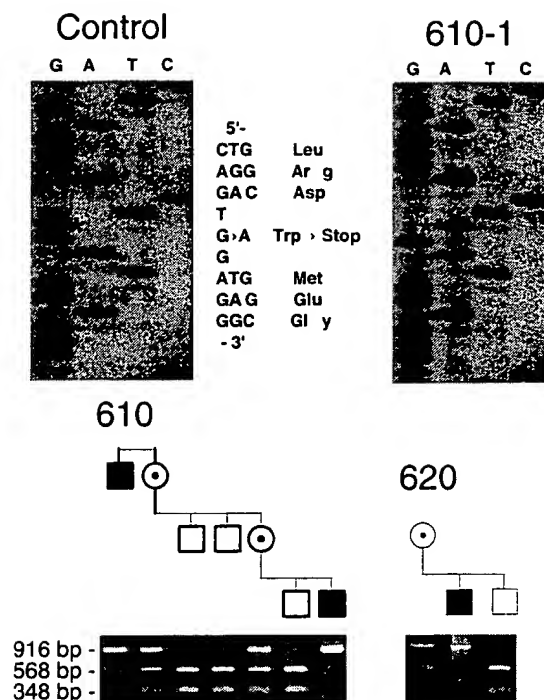
*d*



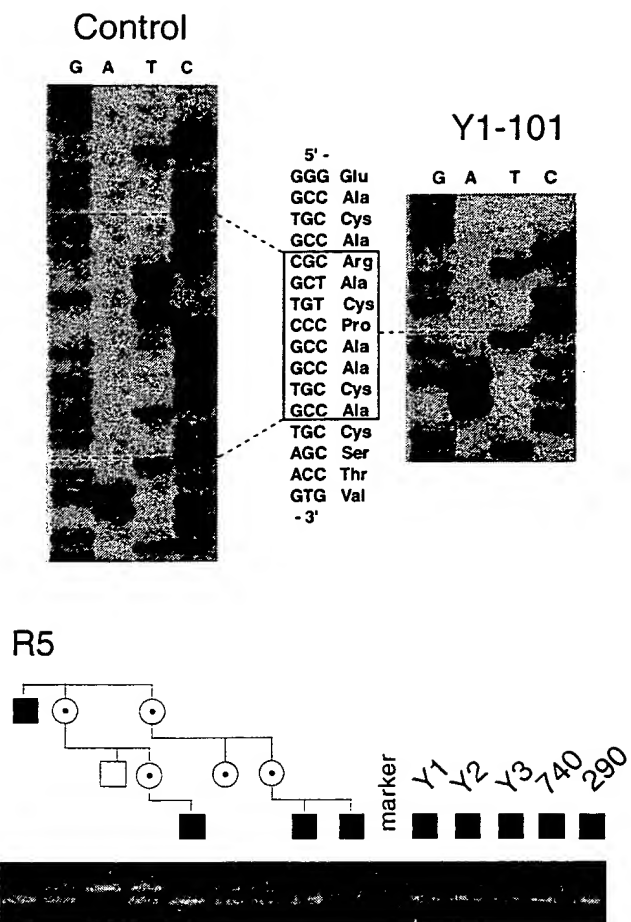
**RHO-AS**

FIGURE 4

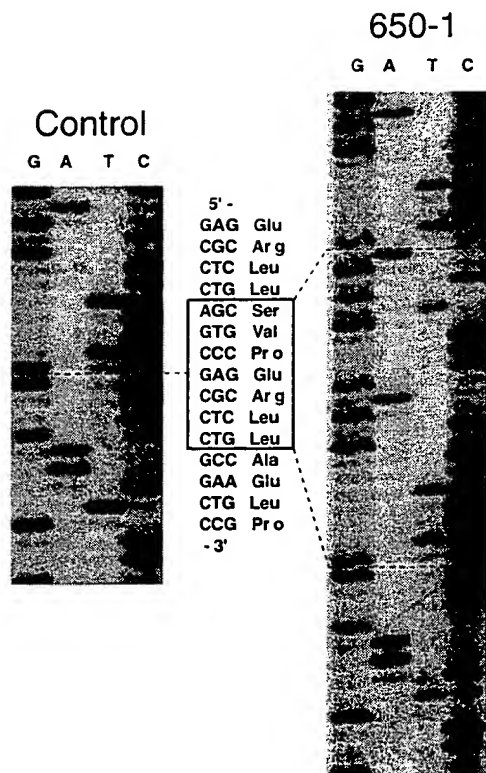
a



b



c



d

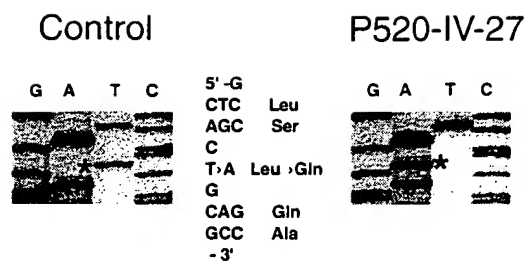


FIGURE 5

Table 1 • Nyctalopin mutations in families with complete CSNB

Family <sup>a</sup>	Origin	Mutation <sup>b</sup>	Codon change	Predicted effect on nyctalopin
290,740,830, Y1, Y2, Y3, R5° USA		85-108del24nt <sup>d</sup>	RACPAACA29-36del	partial loss of N-terminal cysteine cluster
P23.340 (2) (ref 13)	Netherlands	452C>T	P151L	missense, proline to leucine
650 (2)	Canada	464^465ins21nt	SVPERLL155-156ins	expansion of LRR5 <sup>e</sup>
750 <sup>f</sup> (1), 780 (1)	Canada, USA	551T>C	L184P	missense, leucine to proline
540 (6)(ref 28, family 2)	Germany	556-618del50ins3nt		frameshift with stop at codon 259
640 (2)	USA	619^620ins9nt	LLR207-208ins	expansion of LRR6
R7 (7)	USA	628^629ins9nt	CLR209-210ins	expansion of LRR6
P520 (2)	Netherlands	638T>A	L213Q	missense, leucine to glutamine
580 (2)	Canada	647A>G	N216S	missense, asparagine to serine
550 (9)(ref 28, family 3)	Germany	695T>C	L232P	missense, leucine to proline
B1 (3)	USA	792C>G	N264K	missense, asparagine to lysine
B660 (1)	USA	854T>C	L285P	missense, leucine to proline
B2 (5)	USA	893T>C	F298S	missense, phenylalanine to serine
610, 620 (4, 10)	Costa Rica	1049G>A	W350X	protein truncation, loss of GPI-anchoring

<sup>a</sup> families Y1, Y2, Y3, R5, R7, B1 and B2 were first reported in reference 25. Earlier reports on other families is indicated following the family designation.

<sup>b</sup> following the recommendations of Nomenclature Working Group.

<sup>c</sup> number of affected males in these families: 8, 1, 4, 8, 2 and 5, respectively. For all other families in this table this information is

shown in first set of parentheses following the family designation.

<sup>d</sup> in the analysis of X chromosomes with this deletion, we observed identical haplotypes across Xp11.4 from DXS556 to DXS228 with some chromosomes diverging proximally or distally beyond this region (15 markers tested)

<sup>e</sup> LRR, leucine-rich repeat.

<sup>f</sup> Patient 1, whose electrophysiological results are shown in Fig. 1.

FIGURE 6

[illegible]

FIGURE 7